



Frasier syndrome

Frasier syndrome is a condition that affects the kidneys and genitalia.

Frasier syndrome is characterized by kidney disease that begins in early childhood. Affected individuals have a condition called focal segmental glomerulosclerosis, in which scar tissue forms in some glomeruli, which are the tiny blood vessels in the kidneys that filter waste from blood. In people with Frasier syndrome, this condition often leads to kidney failure by adolescence.

Although males with Frasier syndrome have the typical male chromosome pattern (46,XY), they have gonadal dysgenesis, in which external genitalia do not look clearly male or clearly female (ambiguous genitalia) or the genitalia appear completely female. The internal reproductive organs (gonads) are typically undeveloped and referred to as streak gonads. These abnormal gonads are nonfunctional and often become cancerous, so they are usually removed surgically early in life.

Affected females usually have normal genitalia and gonads and have only the kidney features of the condition. Because they do not have all the features of the condition, females are usually given the diagnosis of isolated nephrotic syndrome.

Frequency

Frasier syndrome is thought to be a rare condition; approximately 50 cases have been described in the scientific literature.

Genetic Changes

Mutations in the *WT1* gene cause Frasier syndrome. The *WT1* gene provides instructions for making a protein that regulates the activity of other genes by attaching (binding) to specific regions of DNA. On the basis of this action, the WT1 protein is called a transcription factor. The WT1 protein plays a role in the development of the kidneys and gonads (ovaries in females and testes in males) before birth.

The *WT1* gene mutations that cause Frasier syndrome lead to the production of a protein with an impaired ability to control gene activity and regulate the development of the kidneys and reproductive organs, resulting in the signs and symptoms of Frasier syndrome.

Frasier syndrome has features similar to another condition called Denys-Drash syndrome, which is also caused by mutations in the *WT1* gene. Because these two conditions share a genetic cause and have overlapping features, some researchers have suggested that they are part of a spectrum and not two distinct conditions.

Inheritance Pattern

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Other Names for This Condition

- FS

Diagnosis & Management

These resources address the diagnosis or management of Frasier syndrome:

- Genetic Testing Registry: Frasier syndrome
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0950122/>
- MedlinePlus Encyclopedia: Focal Segmental Glomerulosclerosis
<https://medlineplus.gov/ency/article/000478.htm>
- MedlinePlus Encyclopedia: Nephrotic Syndrome
<https://medlineplus.gov/ency/article/000490.htm>

These resources from MedlinePlus offer information about the diagnosis and management of various health conditions:

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>

Additional Information & Resources

MedlinePlus

- Encyclopedia: Ambiguous Genitalia
<https://medlineplus.gov/ency/article/003269.htm>
- Encyclopedia: Focal Segmental Glomerulosclerosis
<https://medlineplus.gov/ency/article/000478.htm>
- Encyclopedia: Nephrotic Syndrome
<https://medlineplus.gov/ency/article/000490.htm>

- Health Topic: Kidney Diseases
<https://medlineplus.gov/kidneydiseases.html>
- Health Topic: Kidney Failure
<https://medlineplus.gov/kidneyfailure.html>

Genetic and Rare Diseases Information Center

- Frasier syndrome
<https://rarediseases.info.nih.gov/diseases/2375/frasier-syndrome>

Additional NIH Resources

- National Institute of Diabetes and Digestive and Kidney Diseases: Glomerular Diseases
<https://www.niddk.nih.gov/health-information/kidney-disease/glomerular-diseases>

Educational Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology: Frasier Syndrome
<http://atlasgeneticsoncology.org/Kprones/FrasierID10035.html>
- Disease InfoSearch: Frasier Syndrome
<http://www.diseaseinfosearch.org/Frasier+Syndrome/2919>
- Johns Hopkins Medicine: Glomerulosclerosis
http://www.hopkinsmedicine.org/healthlibrary/conditions/adult/kidney_and_urinary_system_disorders/glomerulosclerosis_85,P01475/
- MalaCards: frasier syndrome
http://www.malacards.org/card/frasier_syndrome
- Merck Manual Consumer Version: Nephrotic Syndrome
<http://www.merckmanuals.com/home/kidney-and-urinary-tract-disorders/kidney-filtering-disorders/nephrotic-syndrome>
- My46 Trait Profile
<https://www.my46.org/trait-document?trait=WT1-related%20Wilms%20tumor&type=profile>
- Orphanet: Frasier syndrome
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=347

Patient Support and Advocacy Resources

- March of Dimes: Genital and Urinary Tract Defects
<http://www.marchofdimes.org/baby/genital-and-urinary-tract-defects.aspx>
- National Kidney Foundation: Focal Glomerulosclerosis
<https://www.kidney.org/atoz/content/focal>

- University of Kansas Resource List: Kidney/Urological Conditions
<http://www.kumc.edu/gec/support/kidney.html>
- University of Kansas Resource List: Sexuality and Sexual Differentiation Syndromes
<http://www.kumc.edu/gec/support/ambig.html>

Genetic Testing Registry

- Frasier syndrome
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0950122/>

ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22Frasier+syndrome%22>

Scientific articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28Frasier+syndrome%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- FRASIER SYNDROME
<http://omim.org/entry/136680>

Sources for This Summary

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